Normal human pluripotent stem cell lines exhibit pervasive mosaic aneuploidy.

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Public Summary:
We report the appearance of mosaic aneuploidy in a line of human pluripotent stem cells, that may affect their therapeutic efficacy and safety following transplantation.

Scientific Abstract:
Human pluripotent stem cell (hPSC) lines have been considered to be homogeneously euploid. Here we report that normal hPSC--including induced pluripotent--lines are karyotypic mosaics of euploid cells intermixed with many cells showing non-clonal aneuploidies as identified by chromosome counting, spectral karyotyping (SKY) and fluorescent in situ hybridization (FISH) of interphase/non-mitotic cells. This mosaic aneuploidy resembles that observed in progenitor cells of the developing brain and preimplantation embryos, suggesting that it is a normal, rather than pathological, feature of stem cell lines. The karyotypic heterogeneity generated by mosaic aneuploidy may contribute to the reported functional and phenotypic heterogeneity of hPSCs lines, as well as their therapeutic efficacy and safety following transplantation.